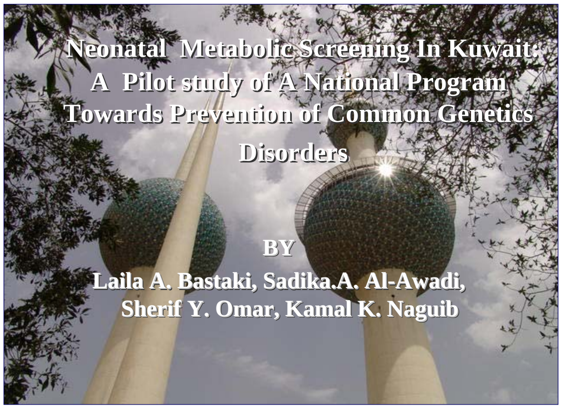

Neonatal Metabolic Screening In Kuwait: A Pilot study of A National Program Towards Prevention of Common Genetics Disorders

BY

Laila A. Bastaki, Sadika.A. Al-Awadi,
Sherif Y. Omar, Kamal K. Naguib

Introduction

- Population in Kuwait: 2,000,000
- Birth rate in Kuwait: 22 per thousand population each year.
- Total annual deliveries: 44000
- Frequency of consanguineous marriages 54.3%

Introduction

- Inherited disease incidence varies from 1:2500 up to 1:20000 live births.
- In Kuwait, the expected incidence will be much higher due to:
  - Personal experience.
  - Large family size.
  - High rate of consanguineous marriage.
  - Genetic isolates.
  - Higher frequency of AR disorders compared to western World.

Introduction

- Previous experience with neonatal screening in Kuwait showed high incidence of:

- Congenital hypothyroidism
(1:3600 live births)

- Phenylketonurea
(1:10000 live births)

Cost Benefit

- The expected annual expenses = 44000 KD
- The expected detected cases of congenital hypothyroidism (12) and phenylketonurea (5)
- The expected total detected cases = 17 detected cases
- The expected cost per case 2588.2 KD

Material and Methods

- Newborn screening occurs in 4 governmental hospitals (Maternity, Farwaniya, Adan & Jahra hospitals).
- Heelstick samples are collected at 3-7 days, sent to the lab within 24 hrs and reported within 3-5 days. Protocol is Delfia for CH and PKU.
- Screening for other inborn errors of metabolism including amino acids, organic acids, fatty acids, carnitine and acylcarnitine disorders was introduced using tandem mass spectrometry in cooperation with Faculty of Pharmacy Kuwait University.

RESULTS

- Total number of newborn screened during period from 1/1/2005 to 31/12/2005 were 3029 cases.
- Total No. of abnormal screening results during this period were 39 cases – 20 CH, 11 HPA, 8 other

- CH (20) - VLCHAD (1)
- Hyperphenylalaninemia (11) - pyruvate carboxylase def (1)
- Tyrosinemia (2) - Non-ketotic hyperglycinemia (1)
- LCHAD (2) - Methylmalonic academia (1)

Discussion

- The incidence of CH exceeds the expected figure by three-fold (8:10,000 vs. 3:10,000)
- The incidence of HPA exceeds the expected figure by four-fold (3.8/10,000).
- This could be due to:
  - False positive results.
  - Transient conditions
  - Early collection of the samples.
  - Frequent heterozygous carriers.
  - Frequent consanguineous marriages.
  - Unexplained situation.
  - Actual figures.

Limitations

- Not all the newborn were subjected to newborn screening (screening only occurred in 4 governmental hospitals and only to newborns in SCU units in these hospitals.
- Problems with sample collection and handling:
  - Time of specimen collection
  - Insufficient blood sampling & poorly saturated filter paper
  - Layering of successive drops of blood in the collection circle
  - Incomplete information on the specimen card.
- Problems with recall and follow up of cases

Recommendations

- Establish a national NBS program.
- Develop a screening protocol for all parts of the screening system:
  - Create a health education campaign
  - Train nurses in proper specimen collection
  - Train additional laboratory staff and develop quality assurance
  - Establish better communicatoin links between the screening staff and follow-up physicians
- Improve management of affected infants
- Systematically evaluate all phases of the program (pre-analytic, analytic and post-analytic) including systematic evaluation of program data
- Consider tandem mass spectrometry to widen the scope of the program
- Consider adding other common metabolic abnormalities


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